BIOARRAY
Genetic Diagnosis

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Consent Bioarray Carrier Panel

My signature at the end of this document indicates that I understood and accepted the information contained herein and that I had the opportunity to clarify all my questions. I give, therefore, consent to Bioarray S.L. to use these samples in the performance of the genetic study indicated in the attached form, as well as in other centers assigned by it when necessary.

WHAT IS THE CARRIER PANEL?

The carrier test developed by Bioarray is a genetic analysis that simultaneously studies, using mass sequencing techniques - NGS, mutations in 420 genes responsible for hereditary recessive pathologies.

Recessive hereditary diseases occur when two mutated copies of the same gene are inherited from the parents. Each copy (allele) is inherited from one of the parents (one from the father and one from the mother) who are carriers.
Patients carrying recessive mutations are healthy patients, in fact, all of us are carriers of some mutations in recessive genes.

If a couple is a carrier of mutations in the same gene, their offspring have a 25% risk of transmitting it and having an affected child, 50% of the offspring are carriers without the disease and another 25% are free of the mutation (non-carriers).
In X-linked recessive diseases, the causative genes are found on the female sex chromosome. Therefore, 50% of the offspring of a carrier mother will receive the mutated copy of the gene. This copy will affect a child differently. Males receiving this mutated copy of the gene will be affected and manifest the disease. In the case of girls, if they have only one mutated copy, they will be carriers but will not have clinical manifestation (although this cannot be completely ruled out due to the phenomenon of X chromosome inactivation).

The availability of this information helps the couple to make appropriate reproductive decisions and, if necessary, to use currently available techniques to reduce reproductive risks and decide on future actions.

With the BIOARRAY recessive disease carriers test we can find mutations with different implications for the patients and their relatives. We can include them in several groups:

- Mutations associated with serious diseases with a significant impact on health that can affect people's life expectancy and quality of life.
- Mutations associated with diseases of varying severity, which can affect to a more variable degree the quality of life of people, but not life expectancy
- Mutations associated with rare diseases, with a significant clinical effect, and which occur with a high incidence in specific populations.
- Mutations present in X-linked genes.

DESCRIPTION OF THE TEST

This test examines a specific number of genes of the patient in search of genetic alterations that help to determine if the patient is a carrier of mutations associated with a series of pathologies. Mass sequencing techniques are used to identify known mutations in specific genome genes. This technique, unlike others, allows us to analyze millions of DNA fragments at a time, allowing us to analyze a large number of genes in a single process.

This test cannot determine all types of mutations; especially, it does not detect alterations that affect genes not included in the test. Your doctor can provide you with information about the specific changes that the test detects.

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ABOUT THE TEST RESULTS

A positive test result indicates that a genetic alteration has been found. A negative result indicates that either no alterations have been found, or the alterations found are of unknown significance (variants that are not known if they are related to a disease).

Although this test detects a high number of mutations, it may not detect, due to technical limitations, the total number of mutations causing recessive diseases.
This test is not the only way to look for genetic alterations, so my doctor may recommend the test before or after other genetic tests.

The results of this test may have implications for me and my family.
It is advisable for the patient/partner to receive genetic counselling before and after the test is performed. Given the complexity of the genetic studies and their important implications, the results will always be communicated to me through a doctor or genetic expert of my choice, and always with the utmost confidentiality.

1. There can be incidental findings: alterations that are detected coincidentally and are unrelated to the disease or condition being studied. However, they can have relevant effects on the patient and/or their family's health.

2. In this analysis, variants of uncertain significance (VUS) can be found. This means that an alteration which has an unknown effect on the pathology has been detected, meaning that it could be a benign variant or the cause of an alteration. In these cases, it could be necessary to analyze the parents to determine whether the alteration is the cause of the pathology or not.

PROCEDURE

To test the DNA in your blood, your doctor will take a blood sample (between 5 and 7 mL, in a standard blood sample). The physical risk to the patient is minimal.

CONSERVATION OF SAMPLES

The remaining DNA will be retained for future validation studies. This will require the written consent of the patient at the end of this document. You have the right to refuse such consent at any time, without giving any explanation and without this having any impact on the quality of the treatment you require.

CONFIDENTIALITY

Only experienced personnel will have access to the blood sample and test information and results. All results will be kept confidential in accordance with applicable laws and guidelines. The results will only be disclosed to your doctor and the requesting health care institution.

Only the requested and authorized tests shall be carried out on the identified blood sample.
The results obtained may be used in scientific publications or presentations, but the identity of all persons studied will not be revealed at any time. The collection of the information obtained is part of a laboratory's standard practice for quality purposes, and is required by laboratory accreditation.

DATA PROTECTION

In accordance with data protection regulations, we provide you with the following treatment information:
Responsible party: BIOARRAY, S.L.
Rights that assist you: access, rectification, portability, deletion, limitation and opposition.
More treatment information: http://bioarray.es/en/

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BIOARRAY S.L. is responsible for the processing of personal data of the Interested Party and informs that these data will be treated in accordance with the provisions of Regulation (EU) 2016/679 of April 27 (GDPR) and Organic Law 3/2018 of 5 December (LOPDGDD), so the following treatment information is provided:

Purposes and legitimation of the treatment

By the legitimate interest of the responsible party (GDPR, Article 6.1.f): maintaining a professional relationship, sending communications, analysing data and publishing scientific and informative articles.

By consent of the interested party (GDPR, article 6.1.a): sending communications, analysing data and publishing scientific and informative articles.

Data retention criteria: will be kept for no longer than necessary to maintain the end of the treatment and when it is no longer necessary for this purpose, they will be eliminated with adequate security measures to guarantee the pseudonymisation of the data or the total destruction thereof.

Communication of the data: the data will not be communicated to third parties, except legal obligation.

I give my consent for the storage and preservation of the samples for possible use in the research on genetic disease and I authorize the transfer of the results of the clinical studies in an anonymous form for the study and pharmacological development, the sending of communications, data analysis and publication of scientific and informative articles:

□ Yes □ No

Date: _____

Informed person (Name and signature): _____

ID number (if available): _____

Physician (Name and signature): _____

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TEST REQUISITION FORM

Carrier Screening / Infertility-related Testing

Requisitioner details

Medical Center / Health Facility | Service/Department | Date

First Name | Family Name | E-mail

Address | City

Province/State | Postal Code | Country | Phone

Patient/Donor details

First Name | Family Name | Gender

Birthdate | Medical Record no. | E-mail

Province/State | Postal Code | Phone

Sample information

Sample type | Extraction method | Extraction date

Remarks

Requested testing (please tick appropriate boxes):

- ☐ Karyotype ●
- ☐ Carrier Screening 420-genes **for donors** (20-genes high-frequency subset reported) ●
- ☐ Carrier Screening 420-genes **for patients** (all genes reported - matchings upon request) ●
- ☐ KIR genotyping ●
- ☐ HLA-C genotyping ●
- ☐ Y chromosome microdeletions (AZF region) ●
- ☐ Sperm FISH analysis (5 chromosomes) ●
- ☐ Thrombophilia Panel (Factor II G20210A; Factor V Leiden G1691A; MTHFR C677T and A1298C) ●
- ☐ Miscarriage testing (Products of Conception) ●

Required sample: ● Lithium heparin tube (green top) ● EDTA tube (purple top) ● Plastic container